



Human Variation Resources at NCBI

- OMIM
- GeneTests/GeneReviews
- dbSNP



Human Variation Resources at NCBI

- OMIM
- GeneTests/GeneReviews
- dbSNP
- dbGaP – database of Genotype and Phenotype



Human Variation Resources at NCBI

- OMIM
- GeneTests/GeneReviews
- dbSNP
- dbGaP – database of Genotype and Phenotype
- Medical Sequencing
- CETT – Genetic Testing Results
- RefSeqGene – standard gene based coordinates
- Putting it together

NCBI

110101

Finding dbGaP

<http://view.ncbi.nlm.nih.gov/dbGaP>

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By Studies By Diseases Advanced Search

Disease	Studies	Variables	Documents	Participants	Type of Study
+ Attention Deficit Disorder with Hyperactivity	1	-	-	-	
+ Bipolar Disorder	1	-	-	-	
+ Cataract	1	-	-	-	
+ Diabetic Nephropathy	1	-	-	-	
+ Macular Degeneration	1	-	-	-	
+ Major Depressive Disorder	1	-	-	-	
- Parkinson Disease	2	-	-	-	
NINDS Parkinsonism Study	-	40	4	1498	Case-control
LEAPS	-	-	-	886	Case-control
+ Psoriasis	1	-	-	-	
+ Psoriatic Arthritis	1	-	-	-	
+ Schizophrenia	1	-	-	-	

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Study: National Eye Institute (NEI) Age-Related Eye Disease Study (AREDS) - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/study.cgi?id=phs000001

Study: National Eye Institute (NEI) Age-Related Ey...

[Research Group](#)

Centrum use and progression of age-related cataract in the Age-Related Eye Disease Study: a propensity score approach. AREDS report No. 21. Ophthalmology. 2006 Aug;113(8):1264-70

Diseases Related to This Study (MESH terms)

- [Macular Degeneration](#)
- [Cataract](#)

Attribution

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 - Emily Y. Chew, MD, Co-Investigator.
 - John Paul SanGiovanni, ScD, Project Officer (2003 to 2007).
 - Natalie Kurinij, PhD, Project Officer (1990 to 2003).
 - Robert Sperduto, MD, Director, Lens Project.
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Study: National Eye Institute (NEI) Age-Related Eye Disease Study (AREDS) - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/study.cgi?id=phs000001

Study: National Eye Institute (NEI) Age-Related Ey...

NCBI dbGaP GENOTYPE and PHENOTYPE AREDS

National Eye Institute (NEI) Age-Related Eye Disease Study (AREDS)

Accession: phs000001.v1.p1

Description

The Age-Related Eye Disease Study (AREDS) was initially designed as a long-term multi-center, prospective study of the clinical course of age-related macular degeneration (AMD) and age-related cataract. In addition to collecting natural history data, AREDS included a clinical trial of high-dose vitamin and mineral supplements for AMD and a clinical trial of high-dose vitamin supplements for cataract. AREDS participants were 55 to 80 years of age at enrollment and had to be free of any illness or condition that would make long-term follow-up or compliance with study medications unlikely or difficult. On the basis of fundus photographs graded by a central reading center, best-corrected visual acuity and ophthalmologic evaluations, over 4,700 participants were enrolled in one of several AMD categories, including persons with no AMD.

The clinical trials for AMD and cataract were conducted concurrently. AREDS participants were followed on the clinical trial for a median time of 6.5 years. Subsequent to the conclusion of the clinical trial, participants were followed for an additional 5 years and natural history data were collected. The AREDS research design is detailed in AREDS Report 1. AREDS Report 8 contains the mainline results from the AMD trial; AREDS Report 9 contains the results of the cataract trial. Blood samples were also collected for genetic research. Genetic samples from 600 AREDS participants were evaluated with a genome-wide scan for inclusion in the dbGaP.

It is hoped that this resource will better help researchers understand two important diseases that affect an aging population. These data may be applied to examination and inference on genetic and genetic-environmental bases for age-related diseases of public health significance and may also help elucidate the clinical course of both conditions, generate hypotheses, and aid in the design of clinical trials of preventive interventions.

[AREDS, The National Eye Institute](#)

[AREDS, The EMMES Corporation](#)

- Subjects: 600

Search Within This Study

Search for:

Associated Analyses

- NEI Age-Related Eye Disease Study (AREDS)
- AMD status

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Associated Documents

- NEI Age-Related Eye Disease Study

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NCBI dbGaP Document NEI Age-Related Eye Disease Study

Chapter 7 EXAMINATION PROCEDURES

7.1 INTRODUCTION

The procedures for carrying out the examinations required in the study are described in this chapter. Required ocular examinations include refraction and visual acuity measurements, intraocular pressure measurement, and ophthalmoscopic examination. General characteristic assessments include measurement of height, weight, and blood pressure and determination of past medical history. Risk factor assessments will require the administration of the food frequency and sunlight exposure questionnaires as well as collection of blood specimens. Procedures for participant identification, masking, distribution and management of the supplementation, adherence assessment, and home visit examination are also described. Procedures for taking photographs of the lens and fundus are described in detail in Chapter 8. The schedule and description of participant visits in Chapter 6 outline the examinations required during each visit.

7.2 REFRACTION AND VISUAL ACUITY

A manifest refraction and visual acuity measurement according to the detailed study protocol must be performed during (a) the Qualifying Visit when the visual acuity score using Chart R is 73 letters or less in at least one eye, (b) the Randomization Visit, (c) Annual Visits, and (d) any Nonannual Visit when the visual acuity score using Chart R has dropped by 10 letters or more compared to the Randomization Visit score for the first time. Participants' pupils should not be dilated at the time of visual acuity testing at any study visit; except they may be dilated during the Qualifying Visit. Pinhole acuity will not be tested as part of AREDS. At the Qualifying Visit, visual acuity may be initially assessed utilizing the participant's current distance glasses. At the Nonannual Visits, visual acuity is initially assessed utilizing the previously obtained manifest refraction. Participants will be asked to read the letters on Chart R only (not Charts 1 or 2), using the equipment described in Section 7.2.1. They will start reading from the top left-most letters--first with the right eye and then with the left eye. A visual acuity score will be calculated as described in Section 7.2.3.3. If at the Qualifying Visit

AREDS: NEI Age-Related Eye Disease Study

AREDS: NEI Age-Related Eye Disease Study

Done

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systolic blood pressure - dbGaP Results - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=search&db=gap&term=systolic%20blood%20pressure&doptcm

systolic blood pressure - dbGaP Results

NEI Age-Related Eye Disease Study (AREDS)		
dias04	Sitting diastolic blood pressure (at follow-up year 4)	phv00000098
NEI Age-Related Eye Disease Study (AREDS)		
dias03	Sitting diastolic blood pressure (at follow-up year 3)	phv00000097
NEI Age-Related Eye Disease Study (AREDS)		
dias00	Sitting diastolic blood pressure (at follow-up year 0)	phv00000096
NEI Age-Related Eye Disease Study (AREDS)		
syst13	Sitting systolic blood pressure (at follow-up year 13)	phv00000095
NEI Age-Related Eye Disease Study (AREDS)		
syst12	Sitting systolic blood pressure (at follow-up year 12)	phv00000094
NEI Age-Related Eye Disease Study (AREDS)		
syst11	Sitting systolic blood pressure (at follow-up year 11)	phv00000093
NEI Age-Related Eye Disease Study (AREDS)		
syst10	Sitting systolic blood pressure (at follow-up year 10)	phv00000092
NEI Age-Related Eye Disease Study (AREDS)		
syst09	Sitting systolic blood pressure (at follow-up year 9)	phv00000091
NEI Age-Related Eye Disease Study (AREDS)		
syst08	Sitting systolic blood pressure (at follow-up year 8)	phv00000090
NEI Age-Related Eye Disease Study (AREDS)		
syst07	Sitting systolic blood pressure (at follow-up year 7)	phv00000089
NEI Age-Related Eye Disease Study (AREDS)		
syst06	Sitting systolic blood pressure (at follow-up year 6)	phv00000088
NEI Age-Related Eye Disease Study (AREDS)		

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BASELINE INTERVIEW — PHASE II (dbGaP ID: phd000020) - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/GetDocument.cgi?id=phd000020#V11

BASELINE INTERVIEW — PHASE II (dbGaP ID: phd...

I would like to take your blood pressure now and again later during this interview.

8. Sitting blood pressure. (Participant must have been seated and quiet for at least 5 minutes prior to the measurement. See Section 7.6 of the Manual of Operations.):

a. Systolic (mmHg) ☒

a. Diastolic(mmHg) ☒

b. Certification number of blood pressure examiner:

9. Have you ever smoked cigarettes for a total of 6 months or more? ☒

☐ no

☐ yes

If no, skip to 10

a. How old were you when you first started smoking?

b. Over your lifetime of smoking, on the average, how many packs per day have you smoked?

☐ ≤ ½ pack

☐ > ½, ≤ 1 pack

☐ > 1, ≤ 2 packs

☐ > 2 packs

c. Do you smoke cigarettes at present?

☐ no

☐ yes

If no, skip to e

d. If you currently smoke, how many cigarettes a day do you smoke?

Skip to 10

e. If you do not smoke currently, how old were you when you last quit smoking?

10. Have you ever smoked cigars, a pipe, or chewed tobacco for a total of 6 months or more?

☐ no

☐ yes

Done

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http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/variable.cgi?id=phv00000094

Variable: syst12

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syst12

Accession: phv00000094.v1.p1

>> [NEI Age-Related Eye Disease Study \(AREDS\)](#) >> [syst12](#)

Description

Sitting systolic blood pressure (at follow-up year 12)

Done

Variable: L_ANKLE - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/variable.cgi?id=phv00021576

Variable: L_ANKLE

NCBI dbGaP GENOTYPE and PHENOTYPE BOSTON UNIVERSITY National Heart Lung and Blood Institute People Science Health

L_ANKLE

Accession: phv00021576.v1.p1

>> [Framingham SHARE](#) >> [Framingham SHARE Ankle Arm BP](#) >> [L_ANKLE](#)

Description

SYSTOLIC BLOOD PRESSURE BY DOPPLER IN LEFT ANKLE

Variable: SYSBP - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/variable.cgi?id=phv00019997

Variable: SYSBP

NCBI dbGaP GENOTYPE and PHENOTYPE GENETIC ASSOCIATION INFORMATION NETWORK GoKinD Genetics of Kidneys in Diabetes Study

SYSBP

Accession: phv00019997.v1.p1

>> [Search for Susceptibility Genes for Diabetic Nephropathy in Type 1 Diabetes](#) >> [SYSBP](#)

Description

Systolic blood pressure (mmHg)

Done

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Recommendations for Blood Pressure Measurement in Humans and Experimental Animals: Part 1: Blood Pressure Measurement in Humans: A Statement for Professionals From the Subcommittee of Professional and Clinical Guidelines of the American Heart Association

Hypertension

2005;45:142-161

Published online before print December 20, 2004, doi: 10.1161/01.HYP.0000150859.47929.8e

AHA Scientific Statement

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Blood pressure measurement

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UK

² Consultant in Anaesthesia
Honorary Reader in Anaesthesia PMS
Plymouth Hospitals NHS Trust
Derriford Hospital
Plymouth
Devon

Variable: syst12 - Windows Internet Explorer

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syst12

Accession: phv000000094.v1.p1

>> NEI Age-Related Eye Disease Study (AREDS) >> syst12

Description

Sitting systolic blood pressure (at follow-up year 12)

Variable: L_ANKLE - Windows Internet Explorer

NCBI dbGaP BOSTON UNIVERSITY National Heart Lung and Blood Institute

L_ANKLE

Accession: phv00021576.v1.p1

>> Framingham SHAPE >> Framingham SHAPE Ankle Arm BP >> L_ANKLE

Description

SYSTOLIC BLOOD PRESSURE BY DOPPLER IN LEFT ANKLE

Variable: SYSBP - Windows Internet Explorer

NCBI dbGaP GoKIND

SYSBP

Accession: phv00019997.v1.p1

>> Search for Susceptibility Genes for Diabetic Nephropathy in Type 1 Diabetes >> SYSBP

Description

Systolic blood pressure (mmHg)

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http://www.ncbi.nlm.nih.gov/SNP/GaP.cgi?rm=plotFrame&test_id=1&chr=1&from=194000000&to=196000000&me

GaP Chromosome Browser

Genotype Summary - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/projects/SNP/GenoFreq.cgi?ald=1&snp=rs7529589

AREDS

$-\log_{10}(\text{uncorrected P-value})$

N/A <2 2-3 3-4 4-5 5-6 6-7 >7

LOD score

N/A <0 0-1 1-2 2-3 >3

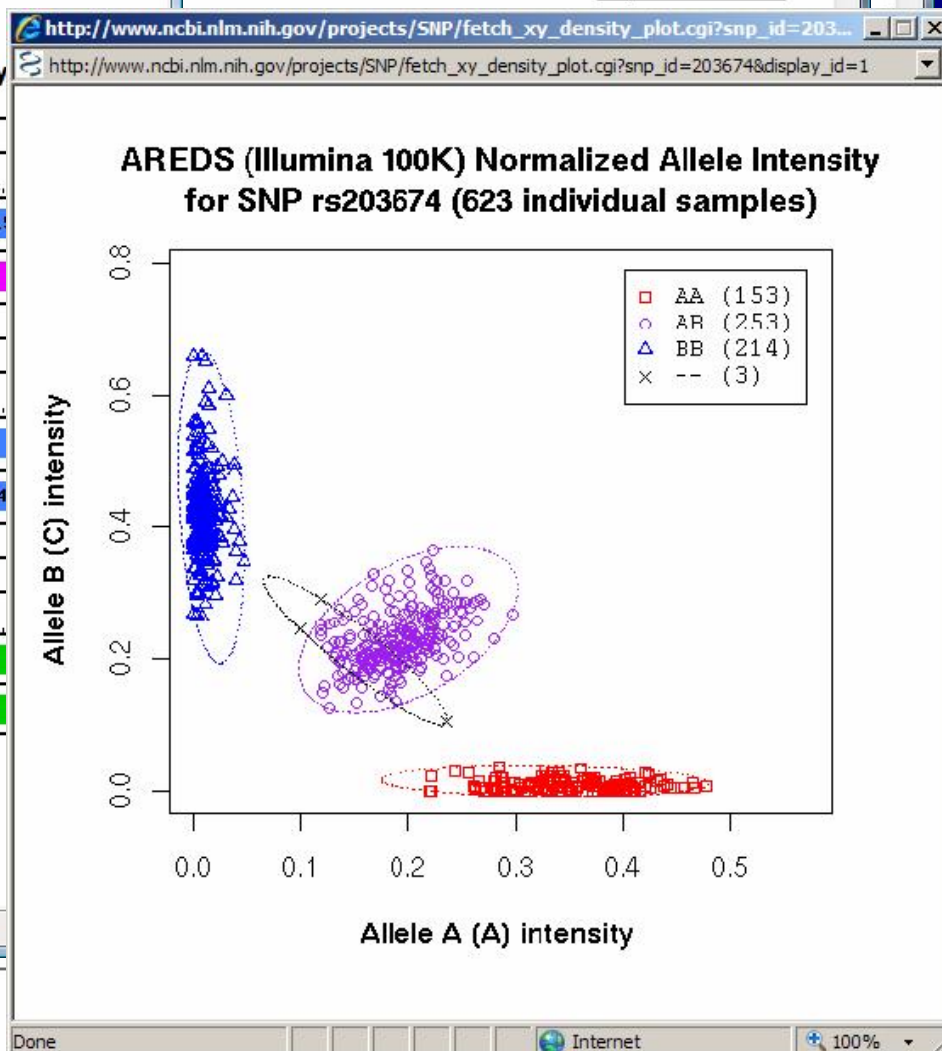
dbGaP study [phs000001](#): rs7529589 Genotype Summary

phv00000173	Genotype			Genotype Frequency		
	CC	CT	TT	25%	50%	75%
Case	63	168	164	15.9%	42.5%	41.6%
Control	80	94	21	41.0%	48.2%	

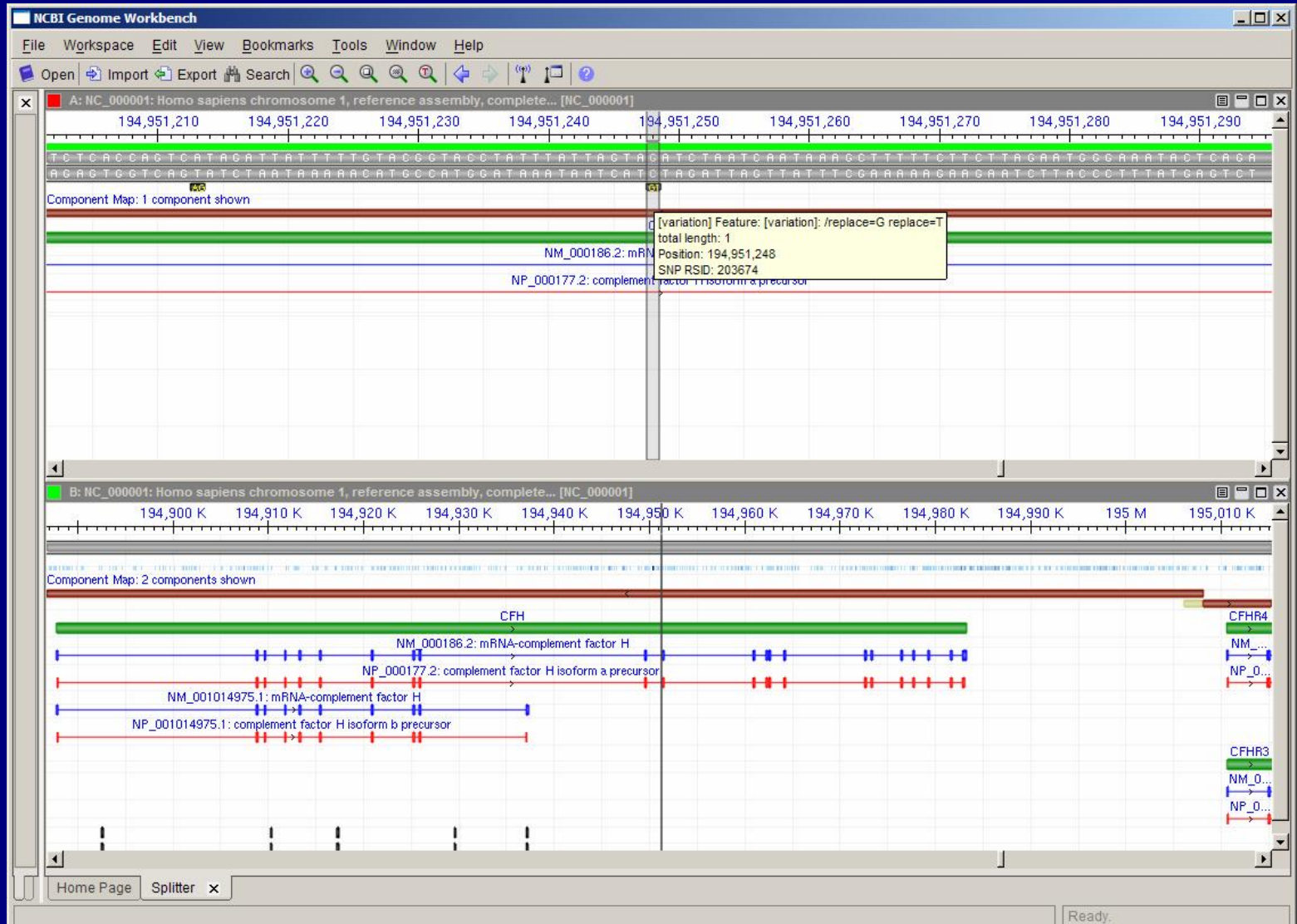
phv00000173	Allele		Allele Frequency		
	C	T	25%	50%	75%
Case	294	496	37.2%	62.8%	
Control	254	136	65.1%	34.9%	

phv00000173	Number of Samples		Success Rate		
	Genotyped	Total	25%	50%	75%
Case	395	395	100.0%		
Control	195	198	98.5%		

Case pHWE: 0.085
 Control pHWE: 0.434
 Odds ratio of minor allele 'C': 0.317
 Chi-square: 80.670
 p-value of Chi-square test: 2.67e-19



Associations to the Basepair

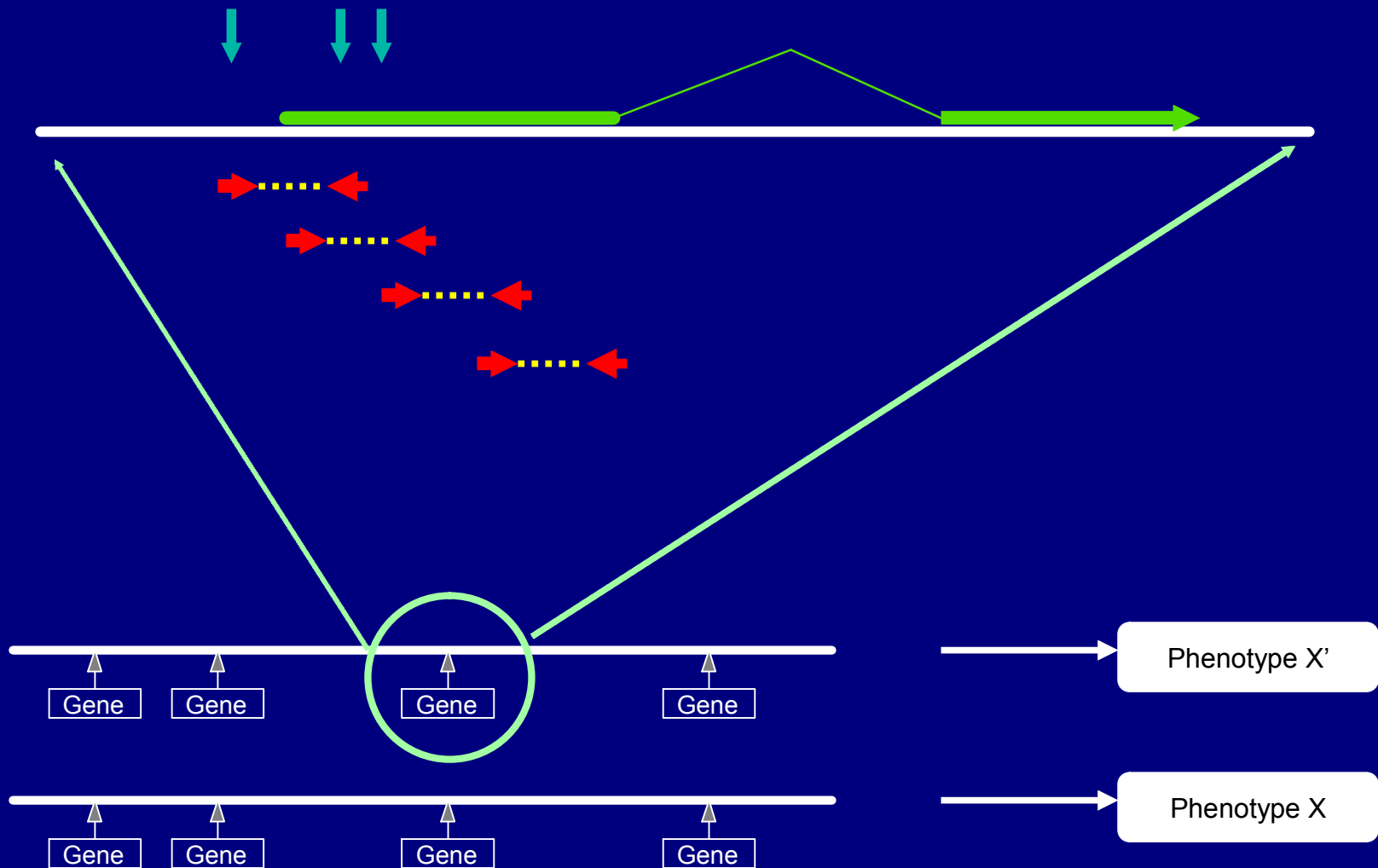




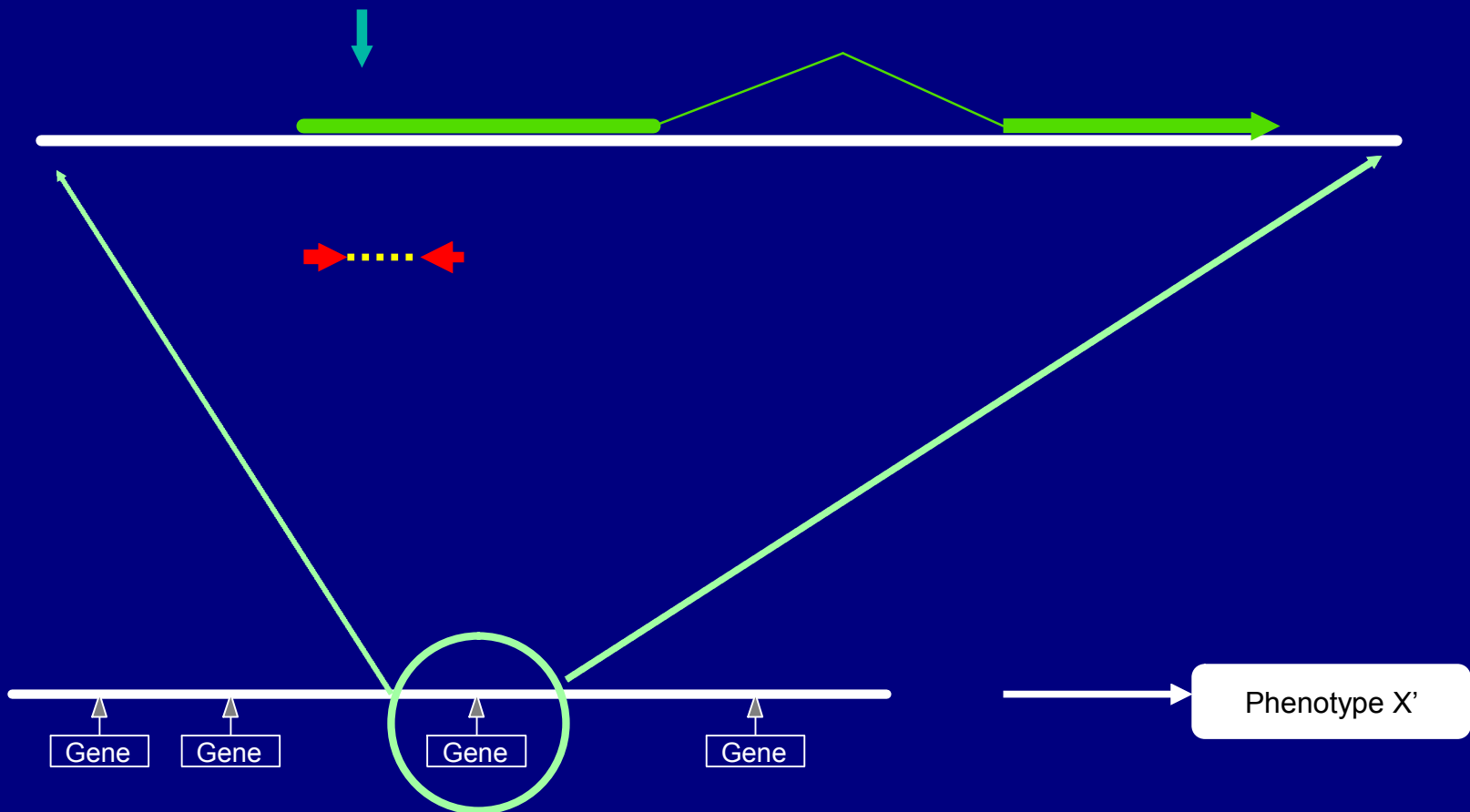
Studies scheduled for dbGaP submission 2007-2008

Projected Availability	Study Name / Disease Focus	Sponsor	Type	Number of Participants
Nov-06	AREDS	NEI	Case-Control GWAS	600
Nov-06	Parkinsonism	NINDS/NIA	Case-Control GWAS	2,573
Jun-07	ADHD	GAIN	Trio GWAS	2,874
Aug-07	Diabetic Nephropathy	GAIN	Case-Control GWAS	1,835
Sep-07	GeneLink	NHLBI	Multipoint linkage analyses	n.d.
Sep-07	Stroke	NINDS	Case-Control GWAS	1,555
Sep-07	Motor Neuron Disease/ALS	NINDS	Case-Control GWAS	1,876
Sep-07	LEAPS	MJFF	Tiered case-control GWAS	886
Sep-07	Major Depression	GAIN	Case-Control GWAS	3,720
Oct-07	Framingham SHARe	NHLBI	Family-Based Longitudinal GWAS	~9,500
Oct-07	Psoriasis	GAIN	Case-Control GWAS	2,898
Nov-07	DCCT/ EDIC	NIDDK	Longitudinal GWAS	
Dec-07	Schizophrenia	GAIN	Case-Control GWAS	2,909
Dec-07	Bipolar Disorder	GAIN	Case-Control GWAS	2,400
Early 2008	Alzheimers	NIA	Case-Control GWAS	10,000
Late 2008	8 GEI Studies	NHGRI	TBD	>30,000
Late 2008	Medical Resequencing, phase 1	NHGRI	TBD	~15,000
Late 2008	MESA SHARe	NHLBI	Longitudinal GWAS	8,000
				99,636

The Medical Sequencing Project – Finding the Causative Mutations



CETT Project – Converting Research to Clinical Tests

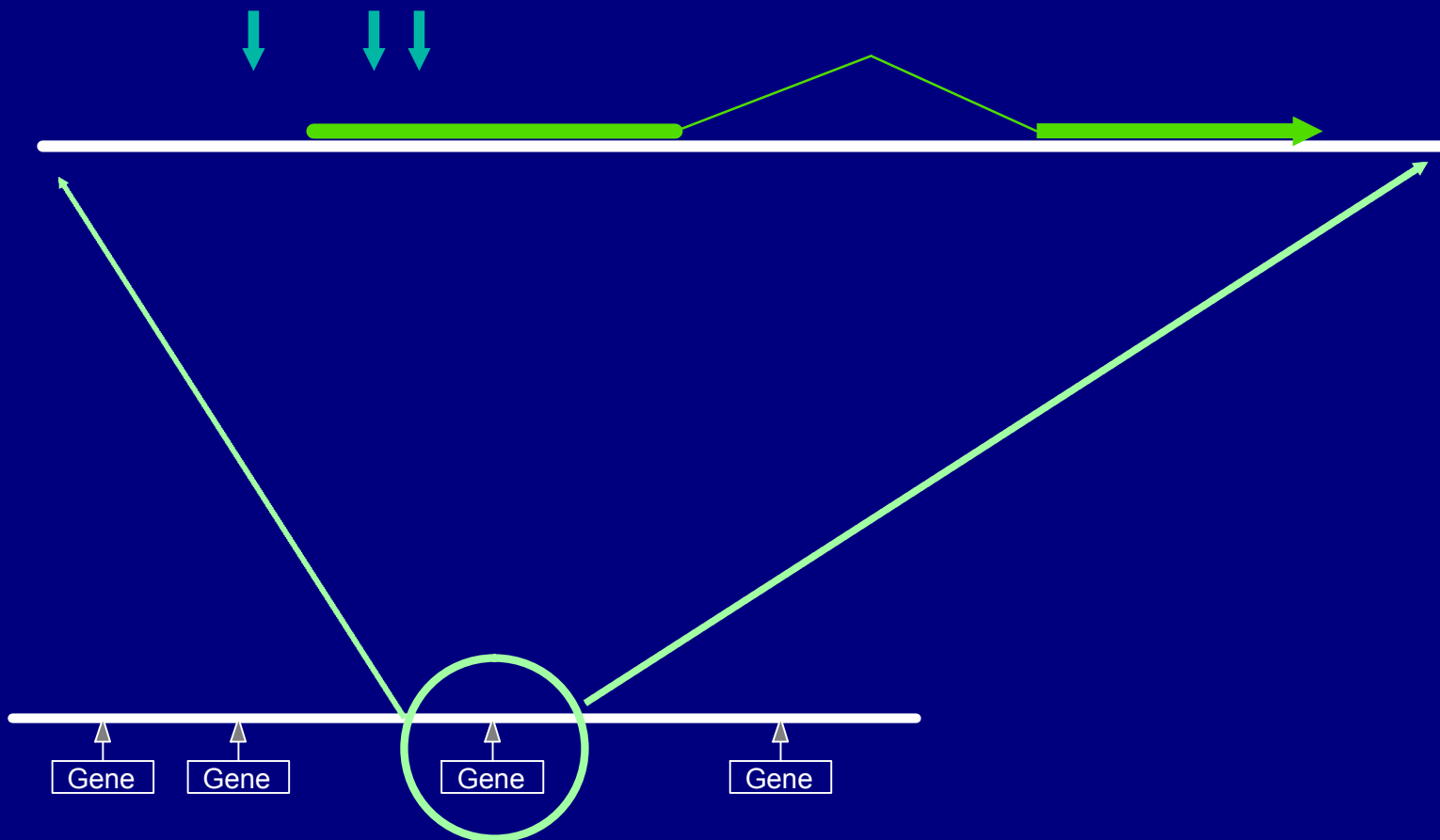


CETT Tests to date

Disease Name	Gene Symbol	OMIM (Gene)	Clinical Laboratory	Yr. 1 # Samples Expected
Infantile Neuroaxonal Dystrophy (INAD)	<u>PLA2G6</u>	603604	OHSU	>100
Kallmann Syndrome (KS)	<u>KAL1</u> <u>FGFR1</u>	308700 136350	GeneDx	>100
Russell Silver Syndrome (RS)	<u>H19</u>	103280	Emory	<50
Progressive Familial Intrahepatic Cholestasis (PFIC)	<u>ATP8B1</u> <u>ABCB11</u> <u>ABCB4</u>	602397 603201 171060	Baylor	50-100
Periventricular Nodular Heterotopia (PVNH)	<u>FLNA</u>	300017	Harvard (Wu)	<50
Xeroderma Pigmentosum (XP)	<u>XPA</u> <u>ERCC3</u> <u>XPC</u> <u>ERCC2</u> <u>DDB2</u> <u>ERCC4</u> <u>ERCC5</u> <u>GTF2H5</u>	278700 133510 278720 278730 278740 278760 122530 608780	UMDNJ/ Harvard (Wu)	<50
Cornelia de Lange Syndrome (CdLS)	<u>NIPBL</u> <u>SMC1L1</u>	608667 300590	U of	60-120 30-60
Autosomal Recessive Agammaglobulinemia (AR-Agama)	<u>IGHM</u> <u>IGLL1</u> <u>CD79A</u> <u>BLNK</u>	147020 146770 112205 604515	Correlagen	25
Arginase Deficiency (Arg Def)	<u>ARG1</u>	608313	UCLA	20-100
MCT8-specific Thyroid Hormone Cell Transporter (THCT) Deficiency (MCT8)	<u>MCT8</u> (<u>SLC16A2</u>)	300095	U of	<12
Galactose Epimerase Deficiency (GALE Def)	<u>GALE</u>	606953	Emory	10
Multiple Acyl-CoA Dehydrogenase Deficiency (Glutaric Acidemia, Type 2) (MADD)	<u>ETFA</u> <u>EFTB</u> <u>EFTDH</u>	608053 130410 231675	U of	~25-50
Deletion 9q34 (9q34)	<u>EHMT1</u>	607001	Emory	20
Mucopolysaccharidosis VI (MPSVI)	<u>ARSB</u>	253200	Emory	~10
Neimann-Pick Disease A and B (NPD A/B)	<u>SMPD1</u>	607608	Emory	~10
Pseudo xanthoma Elasticum (PXE)	<u>A BCC6</u>	603234	GeneDx	300

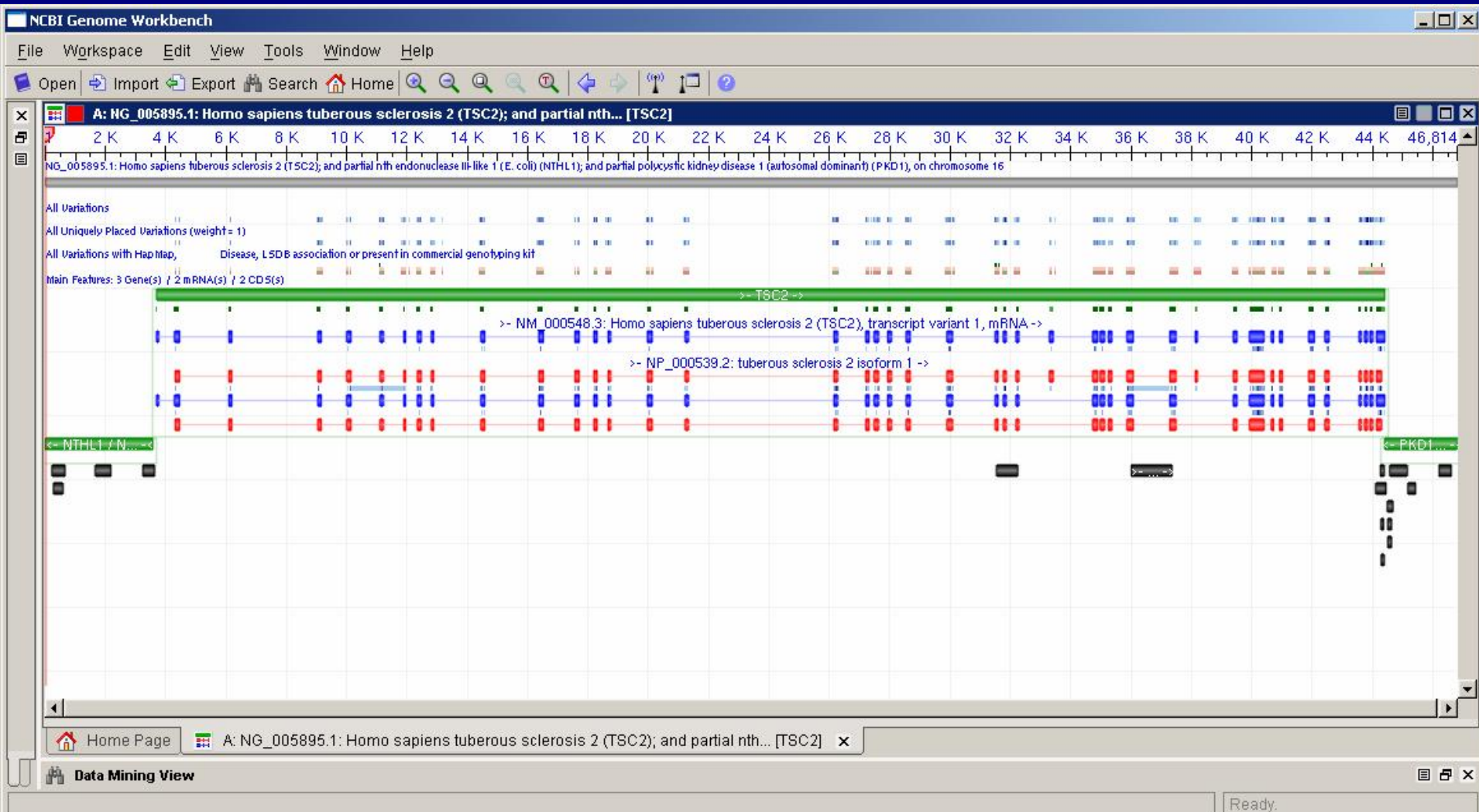
Focal Glomerulosclerosis (Focal GS)	<u>NPHS2</u> <u>4</u> <u>TRPC6</u>	604766 604638 603652	Hosp for Sick Children	38
X-linked Autosomal Recessive Chondrodysplasia Punctata (XLRCDD)	<u>ARSE</u>	300180	GeneDx	50
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)	<u>DSG2</u> <u>DSP</u> <u>PKP2</u>	125671 125647 602861	Hosp for Sick Children	80-380
bilateral frontoparietal polymicrogyria (BFPP) (GPR56 PMG)	<u>GPR56</u>	604110	U of	15-30
Conradi-Hünermann-Happle syndrome, X-linked dominant chondrodysplasia punctata (CDPX2)	<u>EBP</u>	302960	U of	5-10
Cherubism (CRBM)	<u>SH3BP2</u>	118400	Hosp for Sick Children	10-16
Joubert Syndrome (JBTS)	<u>EN1</u> <u>EN2</u> <u>FGF8</u>	131290 131310 600483	Prevention Genetics	<100
Total :23	46		12	

RefSeqGene – Standardizing Clinical Reporting

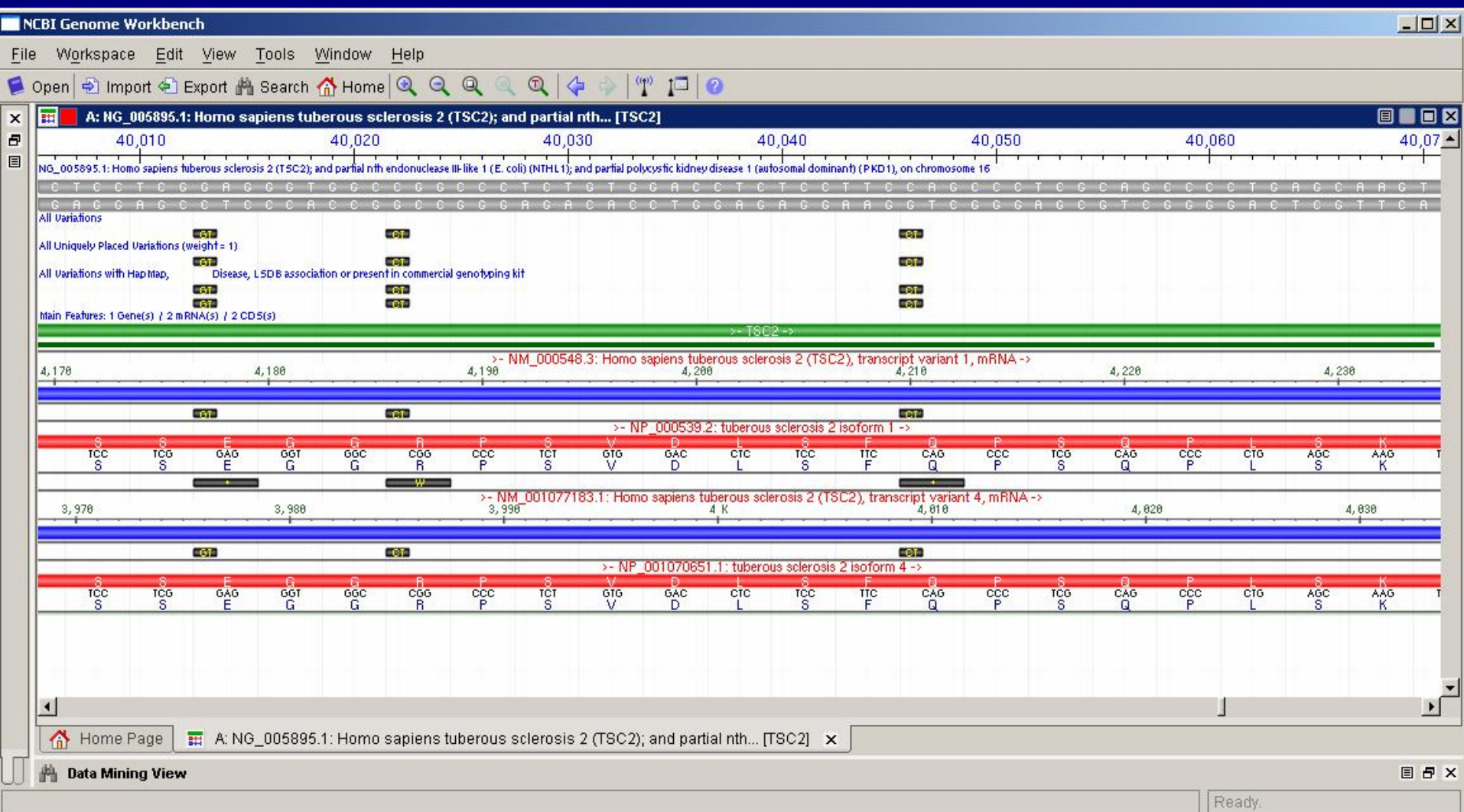




TSC2 RefSeqGene



TSC2 RefSeqGene



TSC2 Variation Report

TSC2 @ chromium.liacs.nl/lovd/ - Windows Internet Explorer

http://chromium.liacs.nl/lovd/search.php?select_db=TSC2&srch=TSC2_00559

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When using or discussing LOVD please refer to:
Fokkema IFAC, Den Dunnen JT and Taschner PEM (2005). LOVD: easy creation of a locus-specific sequence variation database using an "LSDB-in-a-Box" approach. Hum Mutat. 2005 Aug;26(2):63-8.

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Sequence variant tables - Search sequence variants

Total results: 1.

Exon	DNA: Allele 1	RE-Site	RNA	Protein	Frequency	Disease	Reference	DNA/RNA	Technique	TSC2db-ID	Remarks
33	c.4129C>T		r.(?)	p.Gln1377X		TSC	D. Kwiatkowski	DNA	?	TSC2_00559	

Total results: 1. Showing result 1.

Under construction, from [the database of Dr David Kwiatkowski](#), from the published literature and [The Cardiff-Rotterdam Tuberous Sclerosis mutation database](#). The curation of this database is supported by the TSAlliance and it is kindly hosted by the Leiden University Medical Center. The curators are employees of University College London.

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Legend: [[full legend](#)]

Sequence variations are described basically as recommended by the Ad-Hoc Committee for Mutation Nomenclature (AHCMN), with the recently suggested additions (den Dunnen JT and Antonarakis SE [2000], Hum.Mut. 15:7-12); for a summary see [Nomenclature](#), [coding DNA Reference Sequence](#), with the first base of the Met-codon counted as position 1.

Exon: exon numbering. **DNA allele 1:** variation at DNA-level (allele 1). If present, "Full Details" will show you the full-length entry. "Show all records" will show you similar entries. **RE-site:** variation creates (+) or destroys (-) restriction enzyme recognition sequence. **RNA:** variation at RNA-level (allele 1), (?) unknown but probably identical to DNA. **Frequency:** frequency of polymorphism. **Protein:** variation at protein level. **Disease:** disease phenotype, as reported in paper/by submitter, unless modified by the curator (if so, see Remarks column). **Reference:** publication describing the variation, "Submitted:" indicating that the mutation was submitted directly to this database. **DNA/RNA:** variation detected in RNA or DNA. **Technique:** technique used to detect the variation. For a full list of techniques, see the full [legend](#). **TSC2db-ID:** [TSC2 database Identifier](#); if present, links to OMIM ID's are provided. **Remarks:** Listings in bold italics indicate compound heterozygous patients with both mutated alleles known. Consequently, the case is mentioned twice in this table.

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Genotype (Allele 1):

[L48546](#) [NM_000548.2](#) [AB210000.1](#) [AK094152.1](#) [AK125096.1](#)
[NM_000548](#) [NM_021055.1](#) [BC025364.1](#) [BC046929.1](#) [BX647816.1](#)
[NM_021056.1](#) [CR601803.1](#) [CR613640.1](#) [X75621.1](#)

dbMHC locus: [TSC2](#)**Population Diversity**

ss#	Sample Ascertainment				Genotype Detail <small>NEW</small>			Alleles	
	Population	Individual Group	Chrom. Sample Cnt.	Source	C/G	G/G	HWP	C	G
ss11273	HapMap-CEU	European	108	IG	0.019	0.981		0.009	0.991
	HapMap-HCB	Asian	82	IG		1.000			1.000
	HapMap-JPT	Asian	66	IG		1.000			1.000
	HapMap-YRI	Sub-Saharan African	98	IG	0.204	0.796		0.102	0.898

Summary	Average Het. +/- std err:	Individual Count	Founders Count	Individual Overlap	Genotype Conflict
	-1.000+/-0	270	210	0	0

Validation Summary:

[View Genotype](#) [Marker displays](#) [PCR results confirmed](#) [Homozygotes detected](#)

[NCBI TSC2 Resources](#)**Table legend**

dbSNP
rs1748
rs9209
rs30259
rs30260
rs1051616
rs1051621
rs1051758
rs1051771
rs1131826
rs1800705
rs1800712

Submitted Observations of this Variation

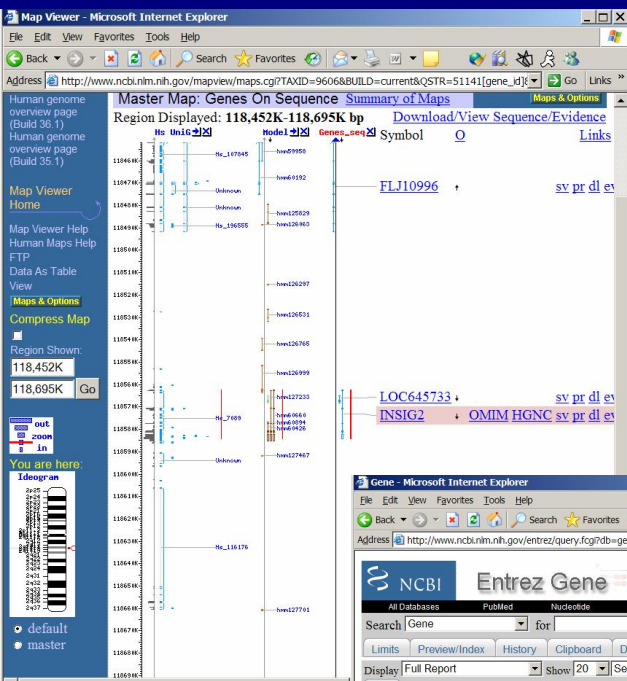
Showing 2 submissions

Method	Submitted by	Link	Details
Computation	Cancer Genome Anatomy Project (CGAP) NCI	[more] 53388	view
Sequence	TSC2 Variation Database University College London	[more] TSC2_00891	view

NCBI



Closing the Loop





Disease InfoSearch

Select Alphabetical
A B C D E F G H I J K L
M N O P Q R S T U V W X
Y Z

What's this?

Organization Search

Select Alphabetical
A B C D E F G H I J K L
M N O P Q R S T U V W X
Y Z

What's this?

Resource Repository

>> Advanced Search
What's this?

Disease

11 Results for

Links provided by

Support Gr

Clinical De

Treatment

General In

Insurance

Reference

See National L

Medication Literature (8886)

Case Studies (3324)

Alternative Medicine Literature (871)

The Hard Science: Genes, Proteins, Etc.

Technical information about the different genes, DNA sequences, chromosomes and protein structures associated with Psoriasis.

Related Genes (20)

[PSORS2](#) : psoriasis susceptibility 2
Homo sapiens , Chromosome: 17 ,
Map Location: 17q , GeneID: 5722

[PSORS4](#) : psoriasis susceptibility 4
Homo sapiens , Chromosome: 1 ,
Map Location: 1cen-q21 ,
GeneID: 10547

[PSORS3](#) : psoriasis susceptibility 3
Homo sapiens , Chromosome: 4 ,
Map Location: 4q , GeneID: 7889

[See all 20 Gene records](#)

Schizophrenia - Windows Internet Explorer

http://web.ncbi.nlm.nih.gov/books/bv.fcgi?rid=huge.chapter.2

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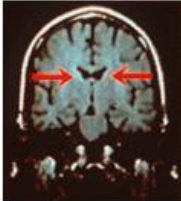
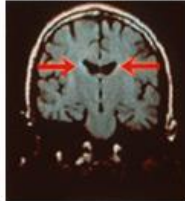
Where are the genes associated with schizophrenia? Check out [MapViewer](#) to see where some of the candidate genes lie in the human genome.

HuGE Encyclopedia

Schizophrenia

About 1 of 100 people develop schizophrenia in their lifetime. Schizophrenia is a psychiatric disorder characterized by delusions and hallucinations. Symptoms often appear in the late teens and multiple relapses throughout adulthood can make it difficult to stay in employment and maintain personal relationships, but medications and support from family members can help.

MRI of Monozygotic Twins (male adults)

No Schizophrenia	Has Schizophrenia
	

The red arrows are pointing to a ventricle (a space filled with cerebral spinal fluid) within the brain. Note how the ventricle is larger in the twin with schizophrenia - this is a common finding in schizophrenia, even when patients are receiving treatment.

There is no clear cut "schizophrenia gene". In fact, over 500 different genes have been linked to schizophrenia. The Schizophrenia Gene Database has whittled down this long list of genomic suspects to 26 different genes. Variations in these genes either decrease (DAO, DRD4, GABRB2, HP, IL1B, PLXNA2, SLC6A4) or increase (APIE, COMT, DRD1, DRD2, DRD4, DTNBP1, GRIN2B, MTHFR, TP53, TPH1) the risk of schizophrenia.

Studies of identical twins show that if one twin has schizophrenia, the other twin is 50% more likely to

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